Kniest dysplasia

Kniest dysplasia is a disorder of bone growth characterized by short stature (dwarfism) with other skeletal abnormalities and problems with vision and hearing.

People with Kniest dysplasia are born with a short trunk and shortened arms and legs. Adult height ranges from 42 inches to 58 inches. Affected individuals have abnormally large joints that can cause pain and restrict movement, limiting physical activity. These joint problems can also lead to arthritis. Other skeletal features may include a rounded upper back that also curves to the side (kyphoscoliosis), severely flattened bones of the spine (platyspondyly), dumbbell-shaped bones in the arms and legs, long and knobby fingers, and an inward- and upward-turning foot (clubfoot).

Individuals with Kniest dysplasia have a round, flat face with bulging and wide-set eyes. Some affected infants are born with an opening in the roof of the mouth called a cleft palate. Infants may also have breathing problems due to weakness of the windpipe. Severe nearsightedness (myopia) and other eye problems are common in Kniest dysplasia. Some eye problems, such as tearing of the back lining of the eye (retinal detachment), can lead to blindness. Hearing loss resulting from recurrent ear infections is also possible.

Frequency

Kniest dysplasia is a rare condition; the exact incidence is unknown.

Genetic Changes

Kniest dysplasia is one of a spectrum of skeletal disorders caused by mutations in the *COL2A1* gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in the clear gel that fills the eyeball (the vitreous) and in cartilage. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears. Type II collagen is essential for the normal development of bones and other connective tissues that form the body's supportive framework.

Most mutations in the *COL2A1* gene that cause Kniest dysplasia interfere with the assembly of type II collagen molecules. Abnormal collagen prevents bones and other connective tissues from developing properly, which leads to the signs and symptoms of Kniest dysplasia.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Kniest chondrodystrophy
- Kniest syndrome
- Metatropic dwarfism, type II
- Metatropic dysplasia type II
- Swiss cheese cartilage dysplasia

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Kniest dysplasia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265279/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Clubfoot
  https://medlineplus.gov/ency/article/001228.htm
- MedlinePlus Encyclopedia: Retinal Detachment
  https://medlineplus.gov/ency/article/001027.htm
- MedlinePlus Encyclopedia: Scoliosis
  https://medlineplus.gov/ency/article/001241.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Clubfoot
  https://medlineplus.gov/ency/article/001228.htm
- Encyclopedia: Retinal Detachment
  https://medlineplus.gov/ency/article/001027.htm
- Encyclopedia: Scoliosis
  https://medlineplus.gov/ency/article/001241.htm
- Health Topic: Bone Diseases
  https://medlineplus.gov/bonediseases.html
- Health Topic: Connective Tissue Disorders
  https://medlineplus.gov/connectivetissuedisorders.html
- Health Topic: Dwarfism
  https://medlineplus.gov/dwarfism.html

Genetic and Rare Diseases Information Center

- Kniest dysplasia
  https://rarediseases.info.nih.gov/diseases/6841/kniest-dysplasia

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases
  https://www.niams.nih.gov/Health_Info/Connective_Tissue/

Educational Resources

- Disease InfoSearch: Kniest dysplasia
  http://www.diseaseinfosearch.org/Kniest+dysplasia/4020
- MalaCards: kniest dysplasia
  http://www.malacards.org/card/kniest_dysplasia
- Nemours Children's Health System
  https://www.nemours.org/service/medical/skeletal-dysplasia/kniest.html?tab=about
- Orphanet: Kniest dysplasia
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=485

Patient Support and Advocacy Resources

- Cleft Palate Foundation
  http://www.cleftline.org
- Human Growth Foundation
  http://hgfound.org/

- International Skeletal Dysplasia Registry, UCLA
  http://ortho.ucla.edu/isdr
- Little People of America
  http://www.lpaonline.org
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/kniest-dysplasia/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/skeldysp.html
- Self Help for Hard of Hearing People (SHHH)
  http://www.hearingloss.org
- The MAGIC Foundation
  https://www.magicfoundation.org/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28kniest+dysplasia%5BTIAB%5D%29+OR+%28kniest+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3240+days%22%5Bdp%5D

OMIM

- KNIEST DYSPLASIA
  http://omim.org/entry/156550

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- Wilkin DJ, Artz AS, South S, Lachman RS, Rimoin DL, Wilcox WR, McKusick VA, Stratakis CA, Francomano CA, Cohn DH. Small deletions in the type II collagen triple helix produce kniest dysplasia. Am J Med Genet. 1999 Jul 16;85(2):105-12.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10406661
- Yokoyama T, Nakatani S, Murakami A. A case of Kniest dysplasia with retinal detachment and the mutation analysis. Am J Ophthalmol. 2003 Dec;136(6):1186-8.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14644246

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/kniest-dysplasia

Reviewed: July 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services